

FIG. 1 RECEIVED

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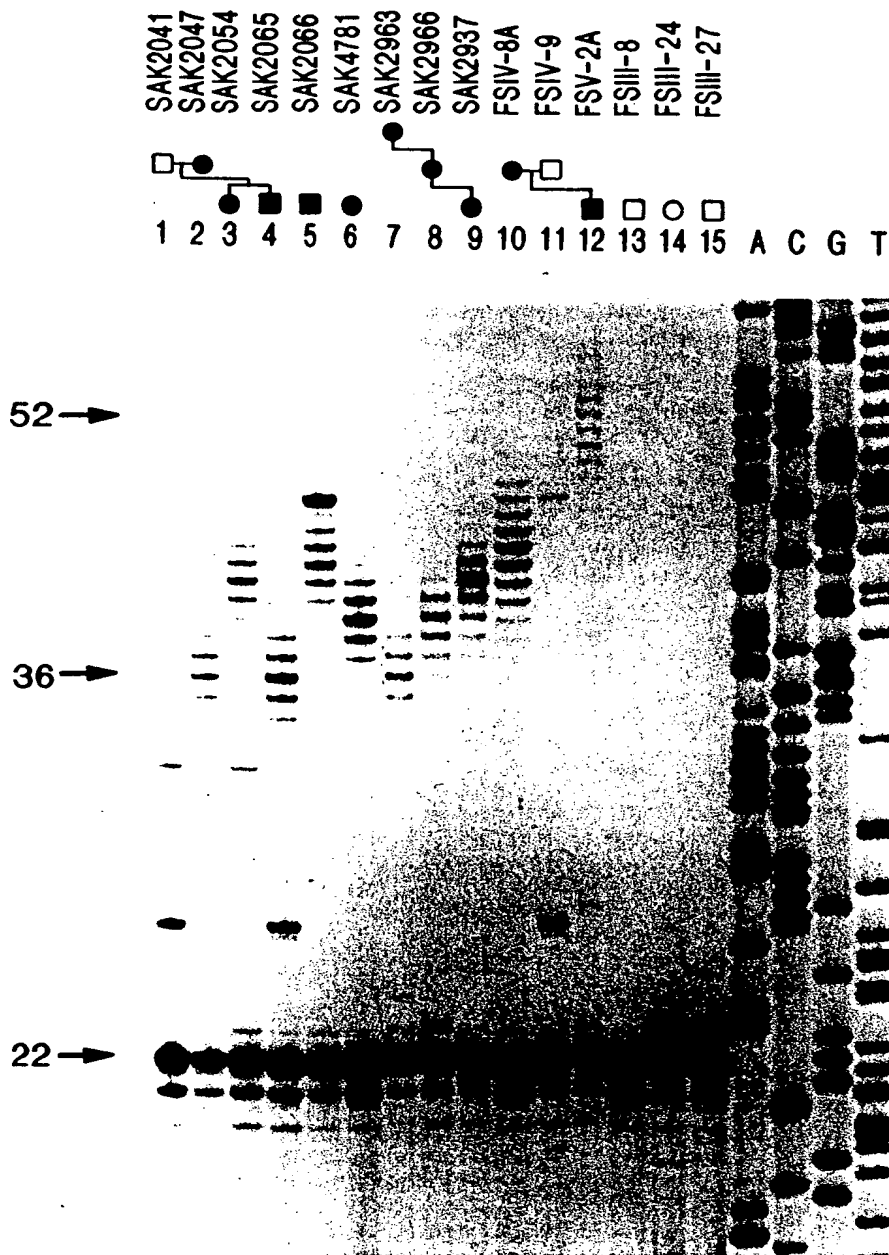
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1 TTGGTAGCAACGGAAACGGCGGGCGGCGGTTTCGGCCCGGCTCCCGGGGGCTCCTTGGTC
61 TCGGCGGGCCTCCCCGCCCCTTCGTCGTCGTCTTCTCCCCCTCGCCAGCCCGGGCGCCC
121 CTCCGGCCGCGCCAACCCGCGCCTCCCCGCTCGGCGCCCGTGCGTCCCGCCGCGTTCCG
181 GCGTCTCCTTGGCGCGCCCGGCTCCC GGCTGTCCCCGCCCGGCGTGCGAGCCGGTGTAT \bar{G}
SCA2-A
241 GGCCCCTCACCATGTCGCTGAAGCCCCAGCAGCAGCAGCAGCAGCAGCAACAGCAGC
SCA2-B
301 AGCAGCAACAGCAGCAGCAGCAGCAGCAGCAGCAGCCGCCGCCGCGGCTGCCAATGTCCGCA
361 AGCCCGGGCGGCAGCGGCCTTCTAGCGTCGCCCGCCGCCGCGCCTTCGCCGTCCTCGTCCT
421 CGGTCTCCTCGTCCTCGGCCACGGCTCCCTCCTCGGTGGTCGCGGGCAGCCTCCGGCGGGCG
481 GGAGGCCCCGGCCTGGGCA \downarrow G GTGGGTGTCGGCACCCC

FIG. 2



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FIG. 3



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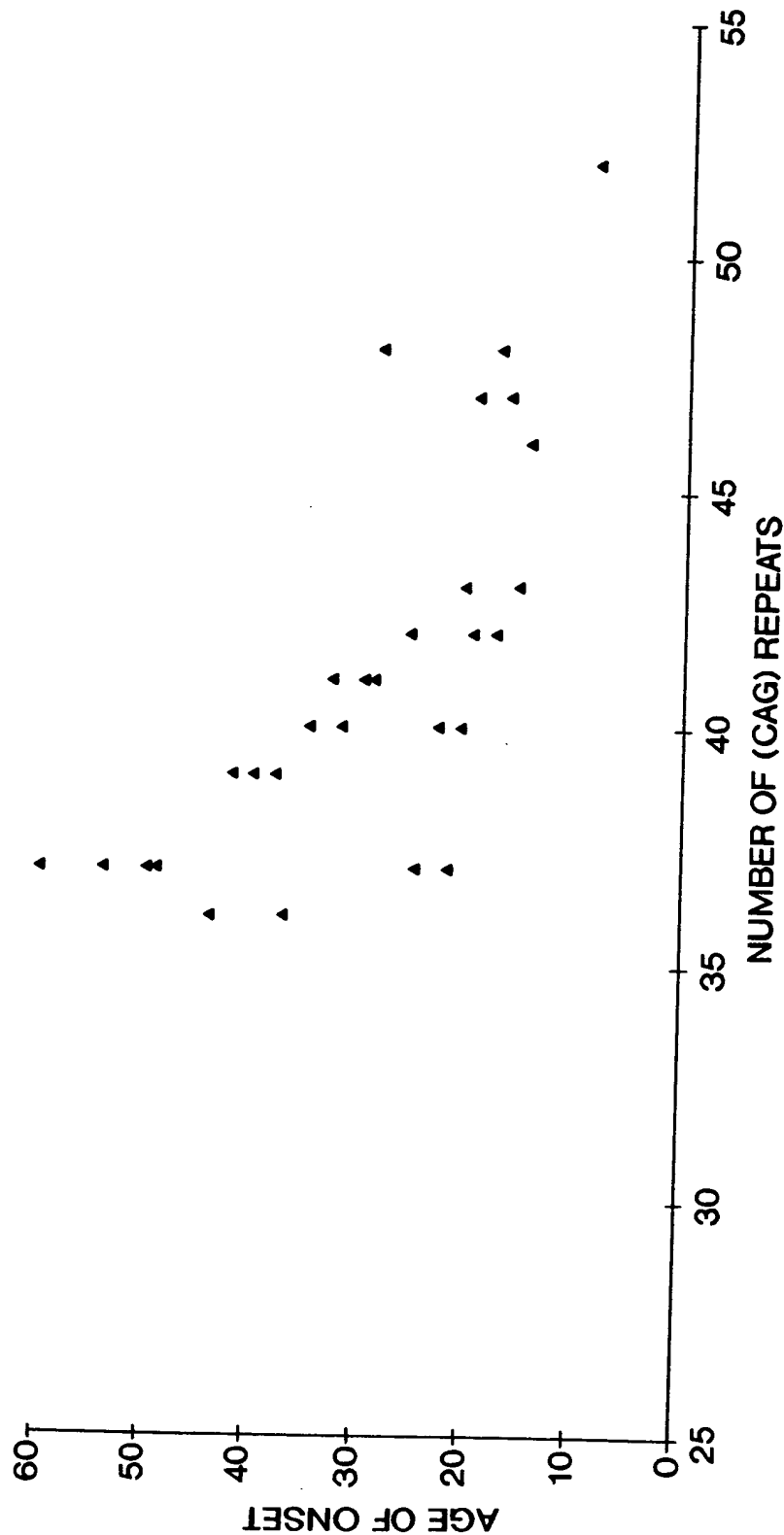


FIG. 4

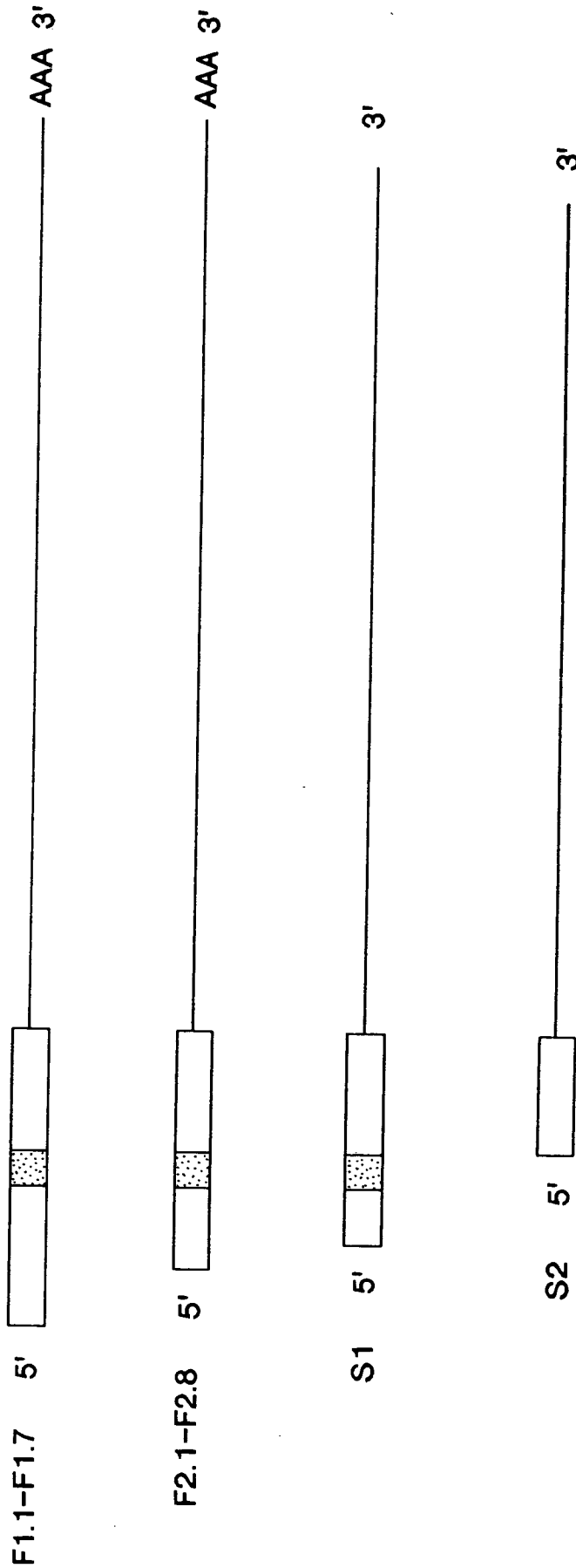


FIG. 5

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FIG. 6B

SCA2-B

721 CAGCCGCGCGCGGTGCCAATGTCCGCAAGCCCGCGGCAGCGGCCTTCTAGCGTCG 780
187 Q P P A A A N V R K P G G S G L L A S 206
781 CCGCGCGCGCGCTTCGCGCTCCTCGTCCCTCGGTCTCCTCGTCCCTCGGCCACGGCTCCC 840
207 P A A A P S P S S S S V S S S A T A P 226
841 TCCTCGGTGTCGCGGACCTCCGCGCGGAGGCCCGCCCTGGGCAGAGGTCGAAAC 900
227 S S V V A A T S G G G R P G L G R G R N 246
901 AGTAACAAAGGACTGCCCTCAGTCTACGATTTCTTTTGATGGAATCTATGCAAAATATGAGG 960
247 S N K G L P Q S T I S F D G I Y A N M R 266
961 ATGGTTCATATACTTACATCAGTTGTTGGCTCCAAATGTGAAGTACAAGTGAAATAATGGA 1020
267 M V H I L T S V V G S K C E V Q V K N G 286

SCA2-14B
1021 GGTATATATGAAGGAGTTTTTAAACTTACAGTCCGAAGTGTGATTTGGTACTTGATGCC 1080
287 G I Y E G V F K T Y S P K C D L V L D A 306
1081 GCACATGAGAAAAGTACAGAAATCCAGTTCGGGGCCGAAACCGTGAAGAAAATAATGGAGAGT 1140
307 A H E K S T E S S S G P K R E E I M E S 326
1141 ATTTTGTTCAAAATGTTTCAGACTTTGTTGTGTACAGTTTAAAGATATGGACTCCAGTTAT 1200
327 I L F K C S D F V V V Q F K D M D S S Y 346
1201 GCAAAAAGAGATGCTTTTACTGACTCTGCTATCAGTGTAAAGTGAATGGCGAACACAAA 1260
347 A K R D A F T D S A I S A K V N G E H K 366
1261 GAGAAGGACCTGGAGCCCTGGGATGCAGGTGAACCTCACAGCCAATGAGGAACCTTGAGGCT 1320
367 E K D L E P W D A G E L T A N E E L E A 386
1321 TTGGAAAATGACGTATCTAATGGATGGGATCCCAATGATATGTTTCGATATAATGAAGAA 1380
387 L E N D V S N G W D P N D M F R Y N E E 406
1381 AATTATGGTGTAGTCTACGTATGATAGCAGTTTATCTTCGTATACAGTGCCCTTAGAA 1440
407 N Y G V V S T Y D S S L S S Y T V P L E 426
1441 AGAGATAACTCAGAAGAATTTTAAACGGGAAGCAAGGCAACCAGTTAGCAGAAGAA 1500
427 R D N S E E F L K R E A R A N Q L A E E 446



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FIG. 6C

1501	ATTGAGTCAAGTCCCCAGTACAAAGCTCGAGTGGCCCTGGAAAATGATGATAGGAGTGAG	1560
447	I E S S A Q Y K A R V A L E N D D R S E	466
1561	GAAGAAAAATACACAGCAGTTCAGAGAAATTCAGTGAACGTGAGGGCACAGCATAAAC	1620
467	E E K Y T A V Q R N S S E R E G H S I N	486
1621	ACTAGGGAAAAATAATATTCTCTCTGGACAAAGAAATAGAGAAGTCATATCTCTGGGA	1680
487	T R E N K Y I P P G Q R N R E V I S W G	506
1681	AGTGGGAGACAGAATTCACCGCGTATGGGCCAGCCTGGATCGGGCTCCATGCCATCAAGA	1740
507	S G R Q N S P R M G Q P G S G S M P S R	526
1741	TCCACTTCTCACACTTCAGATTTCACCCCGAATTCTGTTTCAGACCAAGAGTAGTTAAT	1800
527	S T S H T S D F N P N S G S D Q R V V N	546
1801	GGAGGTGTTCCCTGGCCATCGCCTTGCCCATCTCCTCTCTCGCCACCTTCTCGCTAC	1860
547	G G V P W P S P C P S P S R P P S R Y	566
1861	CAGTCAGGTCCCAACTCTCTCCACCTCGGGCAGCCACCCCTACACGGCCGCCCTCCAGG	1920
567	Q S G P N S L P P R A A T P T R P P S R	586
1921	CCCCCTCGGGCCATCCAGACCCCGTCTCACCCCTCTGCTCATGGTTCTCCAGCTCCT	1980
587	P P S R P S R P P S H P S A H G S P A P	606
1981	GTCTCTACTATGCCCTAAACGCATGTCTTCAGAAAGGCGCTCCAAAGGATGTCCCCAAAGGCC	2040
607	V S T M P P K R M S S E G P P R M S P K A	626
2041	CAGCGACATCCTCGAAATCACAGAGTTTCTGCTGGAGGGTTCCATATCCAGTGGCCTA	2100
627	Q R H P R N H R V S A G R G S I S S G L	646
2101	GAATTTGTATCCACAAACCCACCCAGTGAAGCAGCTACTCCTCCAGTAGCAAGGACCAGT	2160
647	E F V S H N P P S E A A T P P V A R T S	666
2161	CCCTCGGGGGAACGTGGTCATCAGTGGTCAAGTGGGTTCCAAAGATTATCCCCTAAACT	2220
667	P S G G T W S S V V S G V P R L S P K T	686
2221	CATAGACCCAGGTCTCCCAGACAGAACAGTATTGGAATATACCCCCAGTGGGCCAGTTCTT	2280
687	H R P P R S P R Q N S I G N T P S G P V L	706



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FIG. 6D

2281 GCTTCTCCCCAAGCTGGTATTATTCCAACTGAAGCTGTTGCCATGCCCTATTCCAGCTGCA
707 A S P Q A G I I P T E A V A M P I P A A
2341 TCTCCTACGCCCTGCTAGTCTCCTGCATCGAACAGAGCTGTTACCCCTTCTAGTGAGGCTAAA
727 S P T P A S P A S N R A V T P S S E A K
2401 GATTCCAGGCTTCAAGATCAGAGGCAGAACTCTCCTGCAGGGAATAAAGAAAATATTAAA
747 D S R L Q D Q R Q N S P A G N K E N I K
2461 CCCAATGAAACATCACCTAGCTTCTCAAAAGCTGAAAACAAAGGTATATCACCAGTTGTT
767 P N E T S P S F S K A E N K G I S P V V
2521 TCTGAACATAGAAAACAGATTGATGATTAAAGAAAATTTAAGAAATGATTTTAGGTTACAG
787 S E H R K Q I D D L K K F K N D F R L Q
2581 CCAAGTTCTACTTCTGAATCTATGATCAACTACTAAACAAAATAGAGGAGGAGAAAA
807 P S S T S E S M D Q L L N K N R E G E K
2641 TCAAGAGATTTGATCAAAAGACAAAATTGAACCAAGTCTAAGGATTTCTTTCATTGAAAAAT
827 S R D L I K D K I E P S A K D S F I E N
2701 AGCAGCAGCAACTGTACCAGTGGCAGCAGCAAGCCGAATAGCCCCAGCATTTCCCTTCA
847 S S S N C T S G S S K P N S P S I S P S
2761 ATACTTAGTAACACGGAGCACAGAGGGGACCTGAGGTCACCTTCCCAAGGGGTTTCAGACT
867 I L S N T E H K R G P E V T S Q G V Q T
2821 TCCAGCCCAGCATGTAAACAAGAGAAAGACGATAAGGAAGAGAAAGAACGCGCAGCTGAG
887 S S P A C K Q E K D D K E E K K D A A E
2881 CAAGTAGGAAATCAACATTGAATCCCAATGCAAAAGAGTTCAACCCACGTTCTCTCTCT
907 Q V R K S T L N P N A K E F N P R S F S
2941 CAGCCAAAGCCTTCTACTACCCCACTTCACTCGGCCCTCAAGCACAACTAGCCCATCT
927 Q P K P S T T P T S P R P Q A Q P S P S
3001 ATGGTGGTTCATCAACAGCCAACTCCAGTTTATCTACTCAGCCTGTTTGTGACCAAAAT
947 M V G H Q Q P T P V Y T Q P V C F A P N
3061 ATGATGTATCCAGTCCCAGTGAGCCAGCGGTGCAACCTTTATACCCCAATACCTATGACG
967 M M Y P V P V S P G V Q P L Y P I P M T



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3121 CCCATGCCAGTGAATCAAGCCAAGACATATAGAGCAGTACCAAAATATGCCCCCAACAGCGG 3180
987 P M P V N Q A K T Y R A V P N M P Q Q R 1006
3181 CAAGACCAGCATCATCAGAGTGCCATGATGCCACCCAGCGTCAGCAGCGGGCCACCGATT 3240
1007 Q D Q H H Q S A M M H P A S A A G P P I 1026
3241 GCAGCCACCCACCAGCTTACTCCACGCAATATGTTGCCCTACAGTCCTCAGCAGTTCCCA 3300
1027 A A T P P A Y S T Q Y V A Y S P Q Q F P 1046
3301 AATCAGCCCTTGTTCAGCATGTGCCACATTATCAGTCTCAGCATCCTCATGTCTATAGT 3360
1047 N Q P L V Q H V P H Y Q S Q H P H V Y S 1066
3361 CCTGTAATACAGGGTAATGCTAGAAATGATGGCACCACCAACACACGCCCGCTGGTTTA 3420
1067 P V I Q G N A R M A P P T H A Q P G L 1086
3421 GTATCTTCTTCAGCAACTCAGTACGGGCTCATGAGCAGACGATGCGATGTATGCATGT 3480
1087 V S S A T Q Y G A H E Q T H A M Y A C 1106
3481 CCCAAATTACCATAACAAGGAGACAGCCCTTCTTTCTACTTTGCCATTTCACGGGC 3540
1107 P K L P Y N K E T S P S F Y F A I S T G 1126
3541 TCCCTTGCTCAGCAGTATGGCACCCCTAACGCTACCTGCAACCCACATACCTCCACACCT 3600
1127 S L A Q Q Y A H P N A T L H P H T P H P 1146
3601 CAGCCTTCAGCTACCCCACTGGACAGCAGCAAGCCAAACATGGTGAAGTCATCCTGCA 3660
1147 Q P S A T P T G Q Q Q S Q H G G S H P A 1166
3661 CCCAGTCTGTTCAGCACCATCATCAGCACCGCCCGCCAGGCTCTCCATCTGGCCAGTCCA 3720

FIG. 6E



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1167 P S P V Q H H Q H Q A A Q A L H L A S P 1186
3721 CAGCAGCAGTCAGCCATTACACGCGGGCTTGGCCAACTCCACCCTCCATGACACCT 3780
1187 Q Q Q S A I Y H A G L A P T P P S M T P 1206
3781 GCCTCCAAACAGCAGTCGCCACAGAATAGTTCCAGCAGCACACAGACTGTCTTTACG 3840
1207 A S N T Q S P Q N S F P A A Q Q T V F T 1226
3841 ATCCATCCTTCTCAGCCTTACGCGGGGTATACCAACCCACCCACATGGCCACGTACCT 3900
1227 I H P S H V Q P A Y T N P P H M A H V P 1246
3901 CAGGCTCATGTACAGTCAGGAATGGTTCTTCTCATCCAACTGCCCATGGCCCAATGATG 3960
1247 Q A H V Q S G M V P S H P T A H A P M M 1266
3961 CTAATGACGACACAGCCCGGGGTCCCGAGCGCCCTCGCTCAAAGTGCACTACAG 4020
1267 L M T T Q P P G G P Q A A L A Q S A L Q 1286
4021 CCCATTCCAGTCTCGACACACAGCGCATTTCCCTTATATGACGACCCCTTCAGTACAAGCC 4080
1287 P I P V S T T A H F P Y M T H P S V Q A 1306
4081 CACCACCAACAGCAGTTGTAAAGGCTGCCCTGGAGGAACCGAAAGGCCAAATTCCTCCTC 4140
1307 H H Q Q Q L * 1326
4141 CCTTCTACTGCTTCTACCAACTGGAAAGCACAGAAAAC TAGAATTTTCAATTTTGTGTTT 4200
4201 TAAATATATATATGTTGATTTCTTGTAAACATCCAAATAGGAATGCTAACAGTTCACTTGCCAG 4260
4261 TGGAAGATACTTGACCGAGTAGAGGCATTTAGGAACCTTGGGGGCTATTCCATAATTCCA 4320
4321 TATGCTGTTTCAGAGTCCCGCAGGTACCCAGCTCTGCTTGCCGAAACTGGAAGTTATTT 4380
4381 ATTTTAAATAACCTTGAAAGTCATGAACACATCAGCTAGCAAAAAGAAAGTAAACAAGAGT 4440
4441 GATTCTTGCTGCTATTACTGCTAAAAAATAAAAAAAAAA 4481

FIG. 6F



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FIG. 7A

Ataxin-2	1	VYGPLTMSLK	PQQQQQQQQQ	QQQQQQQQQQ	QQQPPAAAN	VRKPGSGLL	50
Mouse Ataxin-2		HEGPLTMSLK	PQPQ		...PPAPAT	GRKPGG.GLL	
A2RP	LA	PQPPPPQHQ	ER	
Consensus		-----L-	PQ	-----	-----	-----	
Ataxin-2	51	ASPAAAPSPS	SSSVSSSSAT	APSSVVA...	ATSGGGRPGL	GRGRNSNKG	100
Mouse Ataxin-2		SSPGAAP.AS	AAVTSASVVP	APAAPVASSS	AAAGGRPGL	GRGRNSSKGL	
A2RP		..PGAAAIGS	A.....RGQSTGKGP	
Consensus		--P-AA---	S	-----	-----	-RG-----KG-	
Ataxin-2	101	PQSTISFDGI	YANMRMVHIL	TSVVGSKCEV	QVKNGGIYEG	VFKTYSPKCD	150
Mouse Ataxin-2		PQPTISFDGI	YANVRMVHIL	TSVVGSKCEV	QVKNGGIYEG	VFKTYSPKCD	
A2RP		PQSPV.FEGV	YNNSRMLHFL	TAVVGSTCDV	KVKNGTTIEG	IFKTLSSKFE	
Consensus		PQ-----F-G-	Y-N-RM-H-L	T-VVGS-C-V	-VKNG--YEG	-FKT-S-K--	
Ataxin-2	151	LVLDAAAEKS	TESSSGPKRE	EIMESILFKC	SDFVVVQFKD	MDSSYAKRDA	200
Mouse Ataxin-2		LVLDAAAEKS	TESSSGPKRE	EIMESVLFKC	SDFVVVQFKD	TDSSYARRDA	
A2RP		LAVDAVHRKA	SEPAGGPRRE	DIVDTMVFKP	SDVMLVHFRN	VDFNYATKDK	
Consensus		L--DA-H-K-	-E---GP-RE	-I-----FK-	SD---V-F--	-D---YA--D-	



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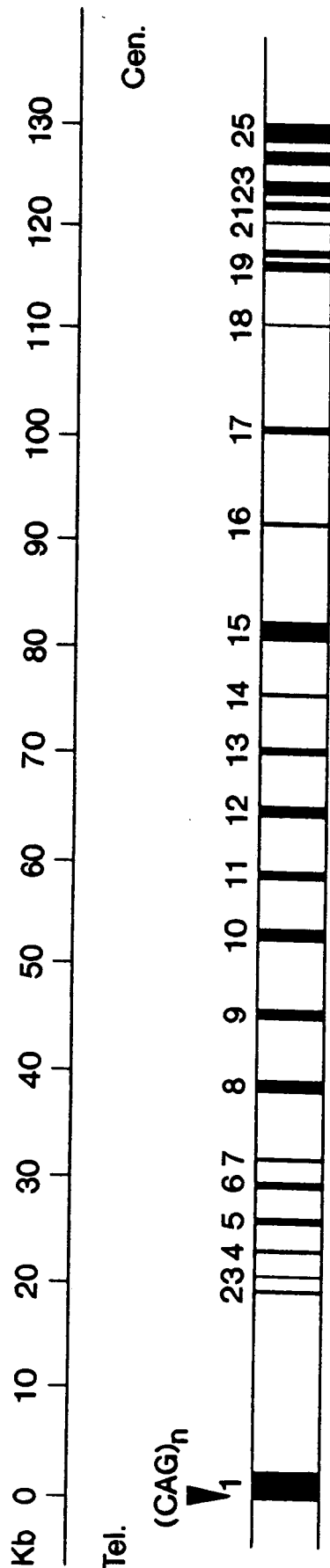
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FIG. 7B

Ataxin-2	201	FTDSAIS..A	KVNGEHKEKD	LEPWDAGELT	ANEELEALEN	DVSNGWDPND	250
Mouse Ataxin-2		FTDSALS..A	KVNGEHKEKD	LEPWDAGELT	ASEELE.LEN	DVSNGWDPND	
A2RP		FTDSAIAMNS	KVNGEHKEKV	LQRWEGGD.S	NSDDYD.LES	DMSGWDPNE	
Consensus		FTDSA-----	KVNGEHKEK-	L--W--G---	-----LE-	D-SNGWDPN-	
Ataxin-2	251	MFRYNEENYG	VVSTYDSSLS	SYTVPLERDN	SEEFKREAR	ANQLAEEIES	300
Mouse Ataxin-2		MFRYNEENYG	VVSTYDSSLS	SYTVPLERDN	SEEFKREAR	ANQLAEEIES	
A2RP		MFKFNEENYG	VKTTYDSSLS	SYTVPLEKDN	SEEFQRELR	AAQLAREIES	
Consensus		MF--NEENYG	V--TYDSSLS	SYTVPLE-DN	SEEF--RE-R	A-QLA-EIES	
Ataxin-2	301	SAQYKARVAL	ENDD.RSEEE	KYTAVQRNSS	EREGHSINTR	ENKYIPPGQR	350
Mouse Ataxin-2		SAQYKARVAL	ENDD.RSEEE	KYTAVQRNCS	DREGHPNTR	DNKYIPPGQR	
A2RP		SPQYRLRIAM	ENDDGRTEEE	KHSAVQRQGS	GRESPLASR	EGKYIP....	
Consensus		S-QY--R-A-	ENDD-R-EEE	K--AVQR--S	-RE-----R	--KYIP----	
Ataxin-2	351	NR					
Mouse Ataxin-2		NR					
A2RP		..					
Consensus		--					

SCA2 Gene



EXONS
Not drawn to scale

INTRONS
Estimated sizes

- Largest exon: exon 1, 928 bps; contains CAG repeat
- Largest intron: intron 1 with approximately 15 Kbps
- Smallest exon: exon 2, 37 bps
- known intron sizes:
 - intron 2 : 1.6 Kb
 - intron 19: 0.3 Kb
 - intron 22: 1.0 Kb
 - intron 24: 1.6 Kb
- Exon sizes:
 - 8<100 bps
 - 100 bps<12<200 bps
 - 200 bps< 4<400 bps
 - 400 bps< 1

FIG. 8

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